

Pink hearing aids and purple shampoo: biographical implications of Waardenburg syndrome type-1

Wheeler, Sara

Auto/Biography Yearbook: The Annual Journal of the British Sociological Association Study Group on Auto/Biography

Published: 17/07/2017

Peer reviewed version

[Cyswllt i'r cyhoeddiad / Link to publication](#)

Dyfyniad o'r fersiwn a gyhoeddwyd / Citation for published version (APA):

Wheeler, S. (2017). Pink hearing aids and purple shampoo: biographical implications of Waardenburg syndrome type-1. *Auto/Biography Yearbook: The Annual Journal of the British Sociological Association Study Group on Auto/Biography*.

Hawliau Cyffredinol / General rights

Copyright and moral rights for the publications made accessible in the public portal are retained by the authors and/or other copyright owners and it is a condition of accessing publications that users recognise and abide by the legal requirements associated with these rights.

- Users may download and print one copy of any publication from the public portal for the purpose of private study or research.
- You may not further distribute the material or use it for any profit-making activity or commercial gain
- You may freely distribute the URL identifying the publication in the public portal ?

Take down policy

If you believe that this document breaches copyright please contact us providing details, and we will remove access to the work immediately and investigate your claim.

Pink hearing aids and purple shampoo: biographical implications of Waardenburg syndrome type-1

Abstract

I have Waardenburg syndrome type 1 (WST1), a rare genetic disorder caused by mutations in the PAX3 gene. It is characterised by varying degrees of sensorineural hearing loss, unusual facial features, and distinctive eye, hair and skin colour – sometimes described as pigmentary abnormalities; these may be present at birth or else manifest during the early life-course. I was born hearing into a Deafhearing family, gradually developing low frequency hearing loss, in itself unusual, made more so by age of onset. My once dark hair has turned prematurely silver and my eye and skin colour have also altered. I exhibit facial characteristics of WST1, notably dystopia canthorum. I received genetic counselling and initially experienced onset of changes as biographical disruption, inkeeping with the predominantly negative framing of WST1. In recent years, however, I have embraced the changes and have begun following their progress with interest. I have incorporated the characteristics into my persona, choosing pretty hearing aids, and using purple shampoo to emphasise my silver hair. In this way, I am treating WST1-related changes as a process of biographical emergence and re-invention, rather than disruption. I present my experiences as an autopathographic case study and narrative of the associated biographical implications.

Introduction

In this paper, I explore my personal experiences of Waardenburg syndrome type 1 (WST1), including the embodied characteristics and changes, and my personal psychosocial and emotional responses. I make the connections between my biography and the wider social, historical and public contexts of the phenomenon; I am thus engaged in classic sociological autobiographical practice, exercising my sociological imagination (Wright Mills 1959). I am also engaged in autoethnographic practice, as I explore my autobiographical account in the context of the adjunct literatures (including medical), and other relevant contextual sources (Sparkes 2017), systematically analysing my experiences to understand the broader cultural experience (Ellis et al. 2011). Additionally, given that my personal account of WST1 might at least in part be seen as an autobiographical medical narrative, I believe it also meets the criterion of an autopathography (Aronson 2000).

According to Aronson (2000), the traditional medical case history is characterized by artificiality, to the detriment of the patient's own narrative. Autopathographies address this deficit by revealing patients' experiences of their ailments. In building a collection of autopathographies, Aronson noted that the subjects covered tend to be "serious, dramatic and fashionable", with cancers and strokes being the most prevalent. Noticing the lack of autopathographies of conditions such as backache, in-growing toenails, renal dialysis, or thyrotoxicosis, Aronson commented:

Perhaps there is room for a book of short individual essays on these and other neglected topics (Aronson 2000, p.1599).

This is an interesting proposal and I feel perhaps my paper answers this call, since WST1 is not one of the 'serious, dramatic and fashionable' conditions identified in the review, yet otherwise shares elements of an (admittedly relatively obscure) chronic condition, and thus might sit well alongside essays about backache and thyrotoxicosis. There are, however, some important ways in which WST1 differs from these kinds of chronic conditions. For example, from certain points of view, many of the key physical characteristics might be seen as merely on the continuum of human variation, rather than emanating from a pathological condition – in much the same way as any other eye, hair and skin colour, and facial characteristics, are viewed. Indeed, Khan (2007) begins her clinical monograph on WS with an anecdote about a student having asked her if there were any genetic syndromes which had only positive characteristics, and that this brought to mind WS. Reflecting on WS, of which there are four distinct types, she addresses WS as a single genetic condition, commenting:

Waardenburg syndrome is a genetic syndrome that can produce moderate to profound sensorineural hearing loss, as well as distinctive eye, hair, and skin colour, and unusual facial features. It differs from other genetic syndromes because the majority of phenotypic features can be concealed, and because some of the phenotypic features such as bright blue eyes are considered physically attractive. Other features, such as a white forelock, are regarded as unusual, but not unattractive, and many people with normal hair colour bleach their hair to obtain such a streak (Khan 2007, p.xi).

Needless to say, I approve of this attitude towards features characteristic of WS, and, without wanting to sound immodest, I would support her assertions by saying

that I personally am generally considered to be moderately attractive, with some people even making positive comments regarding the shape and colour of my eyes, and more recently my hair colour, before becoming aware that they are in any way related to my 'condition'. This then does perhaps problematize somewhat the idea of writing an autopathography of WST1, at least with regards to some of the key, non-sensory features, which might not be considered pathological¹.

However, on initial onset I did find dealing with the depigmentation process, and premature greying of my scalp hair, to be particularly distressing. It was also difficult, mostly during my teenage years, to notice, and have pointed out to me, that I had unusual features – compared with those around me. Thus, whilst these phenotypic characteristics are not necessarily unattractive, they did make me 'different', which is often a difficult position for young people to accept and be comfortable with. Indeed, even with the benefit of hindsight and the confidence, or at least apathy, which comes with maturity, there are still times when being 'different' can be uncomfortable. However, for the most part, I am now comfortable with my physical appearance and with being different and unusual.

On the matter of sensorineural hearing loss, conceptualising this as pathological can also be problematized. Whilst Western biomedical medicine, and the medical models of health and disability, would view hearing loss and deafness as impairments, focussing on how to 'fix' or 'treat' the sensory loss, the social model turns its attention to the social issues which create a disabling environment, for example the barriers caused by the attitudes of people and organisations (Blaxter 2010, pp.10-19). Meanwhile, the cultural model of disability views the social model as unsuitable for describing people who are culturally Deaf (capital D intended) since

Deaf people have their own (signed) languages and cultures, and are thus distinct ethnic groups (Bradbury 2009, p.140).

These models of health and disability provide useful ways of unpacking the concept of hearing loss and societal attitudes towards it. Having done this, the conceptualisation of WST1 as a health condition is further problematized. My own hearing loss has now reached a stage where I have sought treatment in the form of hearing aids. The initial onset of my hearing loss and associated tinnitus, and the realisation of their origins and implications for the future, were the cause of a certain degree of shock and distress for me. However, gaining more knowledge about WST1 and spending time reflecting on my hearing loss and tinnitus, has enabled me to gain a somewhat positive perspective on the matter – including incorporating research about them into my career.

My hearing loss is generally only an issue during interaction with others, and, currently, mostly only if people have quiet, soft and/ or low-frequency voices, or the acoustics of a room are poor. In these situations, provided that the lighting is reasonably good and people do not cover their mouths when they speak, I can often compensate, to a certain extent, with lipreading. However, noisy environments such as crowded rooms are challenging and disorientating, making attempts at communication frustrating. It has become increasingly difficult for me to watch television, however technological advances - including video-streaming on an iPad, have rendered this activity less problematic. Conversely, other activities, such as reading and writing, particularly where higher levels of concentration are required, are somewhat improved by my current level of hearing loss. The associated tinnitus I experience is variable and transient (Wheeler & Hopwood 2015) and also appears

to be irritated by stress, fatigue, and spending prolonged periods of time in noisy environments and/ or concentrating on trying to hear. I have therefore been able to develop strategies to reduce the impact of tinnitus and hearing loss, for example wearing ear plugs in noisy environments, and finding time to spend alone in peaceful surroundings. Thus, whilst there are times when my hearing loss and associated tinnitus can cause disruption and distress, and there are likely to be new challenges to be faced ahead, I have now for the most part been able to accept them and incorporate them into my life.

I am personally not culturally Deaf, not having grown up using a signed language as one of my main forms of communication, nor being a member of Deaf culture or community. However, I have had some contact with, and access to, British Sign Language (BSL) and Deaf culture throughout my life through my Deafhearing family², some members of which are audiotically and culturally Deaf (Baker 2010). In my case, then, WST1 and the associated sensorineural hearing loss, and their status as pathology or not, have at least in part been constructed and influenced by life choices and social forces.

Nevertheless, WST1 is considered, in the medical milieu, to be a health condition. As Khan (2007) points out, 'syndrome' means an aggregate of symptoms or signs associated with a disease process or genetic disorder (p.xi). Thus, whilst I have personally come to view WST1 as a positive part of my identity, I cannot deny that it is generally held to be pathological in nature. There is also a substantial medical literature dedicated to WS. In fact, with regards to WS, the academic literature is almost exclusively from a medical perspective. There are, for example, several case studies through time regarding WS, which have been written from the

medical perspective (Sharma & Arora 2015; Jalilian et al. 2015; Partington 1959). I therefore offer my autopathographic case study to bring the 'patient' perspective to this literature, thus addressing the deficit of patient experience presented by the traditional medical case study (Aronson 2000).

In presenting my case study, I draw on the theoretical literature emanating from Bury's (1982) seminal article, in which he proposed the idea of chronic illness as 'biographical disruption'. This has proved to be a thought provoking sociological concept and although it has been criticised, revisited and revised several times over, by numerous authors, including Bury himself, it has stood the test of time and evolved, providing a conceptual framework for a wider field of biographical experiences. Indeed, in reviewing this literature I noted numerous variations on the theme, such as fracture and flow (Reeve et al. 2010), adjustment and reconstruction (Sveen et al. 2016), abruption and repair (Locock et al. 2009), reinforcement (Carricaburu & Pierret 1995) and impact (Grinyer 2007). I have come to view the literature as a whole to be about 'biographical consequences' of health-related issues. I thus locate my case study within this field of inquiry, sharing my experiences and insights from various stages of the trajectory of WST1.

Waardenburg syndrome type-1

The etymology of Waardenburg syndrome stems from the work of Petrus Johannes Waardenburg, an ophthalmologist and geneticist, who noted that many of his patients had anomalies of the eyebrows, nasal root, iris, and scalp and facial hair, and that these anomalies often appeared alongside deafness (Waardenburg 1951). WS has subsequently been subdivided into four major types WST1-WST4

(Jalilian et al. 2015). As a collective genetic syndrome, these types are characterised by varying degrees of sensorineural hearing loss, distinctive eye, hair and skin colour, and 'unusual' facial features (Khan 2007, p.xi).

In her clinical monograph, Khan (2007) discusses WS types 1 and 3 together (chapter 2), and then types 2 and 4 (chapter 3). Her reason for presenting them in this way is that WS types 1 and 3 are "phenotypically similar" (p13), which is to say the characteristics which can be quantified or described in some way (p5) are similar. Meanwhile, whilst types 2 and 4 have phenotypes similar to types 1 and 3, there are some notable differences, including a lack of dystopia canthorum or musculoskeletal abnormalities, and also the possibility of additional phenotypic features, including ocular albinism (WST2), and Hirschsprung's disease (WST4) (p41). Since I have WST1, I will focus on the phenotypic characteristics associated with this type of WS (and thus also WST3). The phenotypic characteristics of WST1 and WST3 are considered in two subcategories of major (Table 1) and minor (Table 2) characteristics:

Table 1: adapted from Khan 2007, p16-29:

| Major Phenotypic features of WS1 and WS3 | |
|---|--|
| Heterochromia iridis | Different colour irises |
| Bright blue eyes | A distinctive colour |
| Dystopia canthorum | Eyes appear unusually widely spaced; inner corners (canthi) of the eyes are displaced laterally; skin from the nasal bridge produces prominent epicanthi |

| | |
|---|--|
| | folds (skin fold of the upper eyelid, covering the inner corner (medial canthus) of the eye. |
| Depigmented head & facial hair | Loss of pigment (natural colour) – in some individuals this includes a white forelock. |
| Congenital sensorineural hearing loss | Varying degrees and distinct from hearing loss which is a 'normal' part of the ageing process. |
| 1 st degree relative with WS | Parent or sibling also exhibiting symptoms |

Table 2: adapted from Khan 2007, p16-29

| Minor Phenotypic features of WS1 and WS3 | |
|---|---|
| Prematurely grey hair | Scalp hair which has lost its natural colour |
| Congenital leukoderma (Vitiligo) | White patches/ spots on the skin |
| Multiple nevi | Moles, birthmarks, freckles |
| Synophrys | Monobrow |
| Hypoplastic nasal alae | Underdeveloped facial features e.g. sides of the tip of the nose ('wings' or lateral edges) |
| Soft tissue syndactyly of digits | Minor anomalies of the fingers or toes |
| Polydactyly | Extra fingers/ toes |

| | |
|-----------------------|--|
| Mandibular hypoplasia | Facial asymmetry, small lower jaws, temporomandibular joint pain, receding chin |
|-----------------------|--|

During the late 1980s and early 1990s, researchers at the University of Manchester localised the PAX3 gene which causes WS types 1 and 2 (Tassabehji et al. 1993). My family were participants in this study. The genetic mutations which produce WS result from changes in a group of genes called 'homeobox genes' (one of which is the PAX3), which (amongst other things) affect formation and distribution of pigment-producing cells called melanocytes. Melanin is the pigment which provides eye, hair, and skin colour, and is also found in the stria vascularis of the cochlea. Researchers have thus speculated that abnormal migration of melanocytes may be responsible for hearing loss in individuals with WS (Khan 2007, p.9).

The inheritance pattern of WS differs among subtypes. WST1 has an autosomal dominant transmission pattern, which means that there is a 50% chance that a child born to a 'normal parent' and a 'WST1 parent'³ will have WST1 (Khan 2007, p.6). Individuals with WS have an increased risk of having or developing sensorineural hearing losses during their lifetimes (Khan 2007, p.8). A recent systematic review of hearing loss in the four types of WS, found hearing loss prevalence in WST1 to be around 52.3% (Song et al. 2016). This prevalence rate is of course dependent on people like myself with WST1 seeking medical intervention for hearing loss and also then the connection being made, within the medical milieu, between the hearing loss and WS. I self-identified as having WST1. I do not know

whether the possibility of WST1 would have been explored or detected had I not raised the issue myself.

According to Khan (2007), the exact incidence of WS is difficult to calculate because incidence reports are often based on specific populations or ethnic groups (p. 10). This might be further complicated by varying levels of WS between specific populations. For example, on the island of Providencia – Columbia, there is an unusually high frequency of individuals with hearing loss (5 in 1000), with WS accounting for 29% of these cases (Lattig et al. 2008). Whilst Petrus Waardenburg originally estimated that 1 in 42,000 individuals in the general population had WS, researchers in the 1990s estimated that WS occurs in about 1 per 4000 live births (Schaefer 1995 cited in Khan 2007, p.10), and 1 in 10,000 to 20,000 in the general population (Read and Newton 1997, cited in Khan 2007, p.10). WS, as a collection of genetic conditions, are thus considered to be reasonably rare (Song et al. 2016).

My Deafhearing family with WST1

As noted above, my family took part in the research at the University of Manchester where mutations in the PAX3 gene were found to cause WST1 (Tassabehji et al. 1993; Read & Newton 1997). By 'family' here, I refer to intergenerational members of my extended family: myself, my father, my brother, my paternal aunty and my cousin. Since we were all involved in the genetic testing, it has been clinically confirmed that we all carry the mutations of the PAX3 gene and thus have been 'diagnosed' or confirmed as having WST1 (except for my brother, who does not).

However, it is also fairly obvious at first glance that myself, my father, my aunty and my cousin have WST1. We all bear many of the key characteristics. My

aunty and cousin both have the distinctive bright blue eyes and we all exhibit dystopia canthorum, silver hair and various degrees of sensorineural hearing loss; my aunty is d/Deaf, my cousin is profoundly d/Deaf (both having been so since early childhood), my father is hard of hearing and I have low frequency hearing loss. Since we know that the condition emanates from my paternal lineage, and given the highly visual and embodied nature of many of the characteristics, we are able to build a reasonable picture of the history of WST1 throughout our family tree.

My paternal grandfather exhibited many of the facial characteristics of WST1 and his brother Trevor (my great uncle) was d/Deaf. I would need to conduct further research into this side of my family tree to discover more about the likely prevalence of WST1. However, I do have a good quality photograph of my paternal great-grandmother which clearly shows her heterochromia iridis (she had one blue eye and one brown eye) and other WST1 features.

It is very interesting for me to consider this additional dimension in my intergenerational family. I share the current fascination with tracing ancestry through family tree research, which has seen the development of dedicated websites set up to facilitate this kind of research⁵ and programmes such as 'Who do you think you are' (BBC 2016). And as Featherstone et al (2005) note, family research, together with research on health and illness, has long been a core theme of the social sciences in Welsh universities (Featherstone et al. 2005, p.xi)

For me, having a genetic condition, brings an additional dimension of curiosity to exploring my ancestry. It also makes more relevant this particular time and place during which I am doing this research. With the human genome project having been completed between 1990 and 2003, we are living in a time where genomics (the

science of genetics) is a prominent force in western society. Taken alongside the many other technological advances during the same timeframe, our increasing knowledge from genomics opens up new questions about the nature of human existence and experience, and has particular resonance when considering conditions such as WST1 (Shprintzen 2007). Indeed, as Featherstone et al (2005) note:

As genetic medicine increases in prominence and significance, and as more and more disorders are identified as having at least a genetic component, it becomes increasingly pressing that we should understand the social relationships of family and kinship into which genetic risks are, as it were, introduced by the work of geneticists and other professionals (Featherstone et al. 2005, p.1)

Numerous sociological and anthropological themes and questions arise from our increased awareness and knowledge of our genes. Within the family context, these might involve a consideration of what family means in everyday terms and how we feel about, and negotiate, our kinship group. Perhaps more saliently for my current research, is the way in which people conceptualize the familial resemblances and individual differences that are observable among family members. There are also many interesting and important questions which arise from a health professional point of view, which have implications for working with, and treating families such as mine with WST1, including how to communicate information and how this might be understood in the context of everyday understandings of inheritance and kinship. (Featherstone et al. 2005, p.1).

This is perhaps where autopathographies generally have a particularly important role in helping to create a literature which creates a bridge between these

two positions (patient and professional), bringing the patient experiences of particular conditions to the academic literature, drawing on the relevant medical literature in order to situate the lived experiences, helping health professionals to empathise with the positions of their patients (Kearney 2006).

A clear and useful case study of WST1

I exhibit many of the classic characteristics of WST1. I have also, for many years, worked in academic health-related research. This combination has resulted in some interesting scenarios and conversations. For example, when I was in my late twenties I attended a conference to present a poster. I got talking to some other delegates, one of whom worked in genomics. I began talking about WS and she smiled and said that when she had seen me enter the conference venue, from the other side of the room, she had immediately guessed that I had WS. At the time I was a little taken aback and not quite sure how to feel. However, following a rocky trajectory of exploring the embodiment of the condition, I am now very pleased to have WST1 and I am proud of my Deafhearing family, within which the phenotypic characteristics are embraced and appreciated, in terms of family resemblance. I will now very briefly explore my experiences of some of the key features.

Synophrys

The first characteristic I noticed was the 'monobrow'. I was quite young and I don't remember how it came about, but I do recall feeling that monobrows were considered to be unattractive. So I wanted to alter mine to be two separate

eyebrows like other people's. I used a razor. Luckily my mum noticed what I had done and explained that tweezers were probably a better option. I have never looked back and always carry them with me. This is probably the only characteristic which I still struggle to incorporate into my persona. I do not foresee there being a time when I will allow the synophrys to grow back.

Hypoplastic nasal alae

This is a curious characteristic because 'hypoplastic' means 'underdeveloped' and the clinical description of WS noses, compared with 'normal' noses, talks about the tip of the nose (nasal alae) being 'poorly formed' and lacking 'well-developed wings or lateral edges', and that they tend to have 'small nares' (nostrils) (Khan 2007, p.27-28). However, I would assert that this is in fact an attractive feature. I distinctly recall an instance at a school jamboree in primary school where we were involved in a sing-along and they needed someone who had a *trwyn bach smwt* (Welsh for little button nose) and everyone pointed towards me – and I was taken to the front for this part of the song; this was not an unpleasant experience and neither did I feel that it meant my nose was 'underdeveloped'. I still don't.

Dystopia canthorum

This feature is an example of 'beauty in the eye of the beholder'. Described clinically as a 'mid-face anomaly', it results in the eyes appearing unusually widely set, with unusual epicanthal folds of skin around the inner corner of both eyes (Khan

2007, p.22-23). This has, particularly during my childhood, been the subject of some unpleasant name-calling, and I was occasionally made to feel ugly. However, as an adult I have received many compliments regarding my eyes, and I have come to think of my 'unusually shaped/ spaced eyes' as being one of my favourite facial features.

Depigmentation of the eyes, hair and skin (including heterochromia iridis)

The onset of depigmentation during puberty was quite confusing for me as I firstly began to notice silver strands of hair. Thinking that this might have been caused by having my hair permed and the use of styling products, I resolved to cease the use of products immediately, allowing my hair to return to its natural state. I also began removing the silver hairs with tweezers; this developed into what could be termed 'trichotillomania' or 'hair-pulling disorder' (American Psychiatric Association 2013, p.251) which to a certain extent still ails me today. At some point I mentioned the issue to my mum and she explained about the WS and our involvement in the research (I did not remember it). Once I realised the implications of WS I was very upset and I would say that I experienced a 'biographical disruption' as conceptualised by Bury (1982).

Until that moment I did not have any reason to consider myself to be the bearer of a 'syndrome'. Suddenly my self-concept and my perceived biography were called into question and I did indeed experience a 'What is going on here' moment and the subsequent attention to my embodiment of the condition (Bury 1982). I think the trichotillomania could be seen as a symptom of this, as could subsequent

behaviour, including numerous disastrous attempts to dye my hair to conceal the silver colour, tan my skin (to the point of burning), and examining my eyes for signs of increasing heterochromia iridis.

Over the years I would say that I have experienced various biographical implications of WST1, including a pendula reconstruction of self and identity, swinging back and forth between pathological and non-pathological aspects of the self (Yoshida 1993; Homma et al. 2016), particularly in relation to depigmentation, as I have swung back and forth between 'owning the silver' and seeking to disguise it. It was interesting to note that, whilst I am now (mostly) comfortable with my WST1, revisiting this period of initial onset for the purposes of this research, and especially presenting it at a conference event, I was struck with a wave of emotion relating to the biographical disruption, which had, until that point, been largely forgotten.

Interestingly, there is possibly a gendered element to my experiences regarding my hair, in that women whose hair turns grey, regardless of stage in their lives, are expected to maintain youthful hair colour, through dying their hair if necessary. This is true of many similar physical attributes, as Greer (2007) notes:

What is pathological behaviour in a man is required of a woman. A bald man who wears a wig is a ridiculous figure; a bald woman who refuses to wear a wig is being stropky and confrontational (Greer 2007, p.24).

I would argue that this is also true of hair colour. My decision to stick with my natural colour has met with bafflement and even overt disapproval by a variety of people. However, I am now proud of my naturally 'Arctic blonde' hair, in all of its normal-defying glory, and I use a purple shampoo to emphasise its silvery hues.

Also, somewhat bizarrely, silver hair appears to have become fashionable recently, with many celebrities and their followers dying their hair to achieve it. Whilst baffling, I welcome this trend since it means that purple shampoo can now be found in almost every chemist, having previously been quite difficult to come by.

Congenital sensorineural hearing loss (and associated tinnitus)

The last aspects of WST1 that I addressed were the hearing loss and associated tinnitus. Strangely, though these are probably the most normal or doctorable (Heritage & Robinson 2006) of the symptoms, these were the most difficult for me to face. I recall a hearing test at school being the source of much anxiety and feeling that I wanted to somehow get out of having the test; I do not recall the test itself or the outcome. It is odd now to consider that I didn't make the connection between my hearing loss and the deafness experienced by other members of my family. Even after learning about the WS during my teenage years, I still did not turn my attention to my hearing or tinnitus.

Having completed a social science degree, I began to take a professional research interest in WST1. I obtained information from the University of Manchester and went for genetic counselling. As I reached my thirties, my hearing became noticeably worse, as did the tinnitus which, upon reflection, I realised had been in the background since I was a small child (Wheeler & Hopwood 2015). The problem with my low-frequency hearing loss is that I appear to have compensated – lip-reading, and guessing, based on the words I heard. However, having observed my father's trajectory of hearing loss and tinnitus, I decided to take steps to address it sooner rather than later, beginning with getting used to hearing aids.

There is currently an unfortunate turn in hearing-aid rhetoric, towards their concealment rather than accessorising and enjoyment (Withey 2016). This has included a definite change of direction of the hearing aid companies towards producing products in dull colours to match skin and hair (though curiously they are not made in white). Thus, the hearing aids now available on the NHS do not include fun, bright colours as they used to. However, I was lucky enough to initially get some bright pink ones, and then my current ones which are soft pink and white, which I like. I do not wish my hearing aids to blend in or be hidden, in fact I would very much like to have changeable cases so that I could match their colour to my outfits and to suit every occasion. Sadly, this does not appear to be the way things are heading, however perhaps there is some action, patient/ customer-led research to be done here. Meanwhile, I have been enjoying conducting research in the fascinating area of hearing loss and d/Deaf studies, and I have begun to embrace my Deafhearing family heritage, taking part in a sign-singing band with my aunty (Wheeler 2012).

A seriously confronting introspective question

'There is currently no treatment or cure for Waardenburg syndrome'
(Wikipedia 2016).

This sentence is matter-of-factly stated on the Wikipedia page regarding WS and expresses sentiments inkeeping with the way in which 'health conditions' are generally discussed. However, coming across this sentence during the course of my research was quite confronting - a stark reminder of the relative 'abnormality' of my

genetic make-up. Moreover, this sentence poses the interesting, theoretical question: if I could be 'cured' of WST1, would I choose to be?

As I have noted above, I have previously sought to cosmetically modify aspects of my appearance relating to WST1, including 'tweezing' my 'synophrys' to produce two distinct eyebrows, and dying my hair as the depigmentation process began to alter it from brunette to silver⁵. I have also sought 'treatment' for my increasing hearing loss through available technologies, including hearing aids. However, one might now argue that I have rejected the ideas of 'treatment' and 'cure', in favour of 'embracement'. I now enhance my silver locks using purple shampoo and enjoy the accessorising quality of my pink hearing aids. However most importantly, perhaps, is that I no longer feel the need to disguise or hide the fact that I have WST1. It no longer distresses me to recognise the fact that I differ from the central line of genetic 'normality'. The WST1 is a part of me and is an important aspect of my identity; I would thus not wish to be 'cured'.

Conclusion

I feel fortunate to be living in a time and place, and working in a field, where I can explore my genetic difference, with all of its biographical implications, without fear of negative consequences or retribution. However, looking at how I might have been treated in the (not too distant) past, and some current attitudes towards difference in the present, I worry what the future holds for individuals with WST1.

As is often the way, conducting this research has generated as many questions as it has answers, and the topic of WST1 has proved too large to provide

more than a brief overview of my experiences within the confines of this article. However, writing this paper has enabled me to create a good foundation upon which to conduct future research on this topic. I look forward to exploring each aspect of WST1 further, and I encourage others with autobiographical medical narratives, to write about them, thus enriching the autopathographic literature, which is such a valuable resource for health professionals and lay audiences alike.

Notes

1. I do have some other health issues which may form the basis of future autopathographies, however they are, to my knowledge, unrelated to WST1.
2. A term first coined by Donna West during her Master's research, and later developed in her book (see bibliography of this paper) which encapsulates the experience of family life where deafness is not solely attributed to one or two individuals, as a problem to be managed and medicalised, but is rather a shared experience to be pondered, explored and even celebrated.
3. Emphasis using single inverted commas my own (not in the original text).
4. See for example: www.ancestry.co.uk/FamilyTree; www.findingmypast.co.uk/FamilyTree; www.genesreunited.co.uk and many more.
5. In the interests of full disclosure and transparency, I should state that I had rhinoplasty surgery, aged 16. However, the surgery I had was reconstructive as I had broken my nose on several occasions engaging in contact sports such as kung fu and rugby union; my nose was restored to a state resembling

its pre-breaking, rather than modifying my looks away from those associated with WS.

Acknowledgements

I would like to thank everyone who attended the session of the BSA Auto/ Biography Summer Conference 2016, at which I presented the first draft of this paper; I thank you for your interest, encouragement and patience, particularly as I stumbled through some unexpectedly emotional aspects of the narrative.

Author Biography

I am a Lecturer in Social Policy (Welsh medium) at Bangor University, Wales, funded by Y Coleg Cymraeg Cenedlaethol (the National Welsh-medium College).

Correspondence: s.wheeler@bangor.ac.uk

References

- Adamson, J. & Donovan, J., 2005. "Normal disruption": South Asian and African/Caribbean relatives caring for an older family member in the UK. *Social science & medicine*, 60(1), pp.37–48.
- American Psychiatric Association, 2013. *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5)*, Washington, DC: American Psychiatric Publishing.
- Aronson, J.K., 2000. Autopathography: The patient's tale. *British Medical Journal*, 321, pp.1599–1602.

- Baker, C., 2010. Sign Language and the Deaf Community. In J. A. Fishman. & O. García, eds. *Handbook of language and ethnic identity: disciplinary and regional perspectives, Volume 1*. New York: Oxford University Press, pp. 153–171.
- BBC, 2016. Who do you think you are? *BBC one*. Available at: <http://www.bbc.co.uk/programmes/b007t575> [Accessed December 30, 2016].
- Blaxter, M., 2010. *Health*, Cambridge, UK: Polity.
- Bradbury, H., 2009. *Medical Sociology - an introduction*, Los Angeles: SAGE Publications.
- Bury, M., 1982. Chronic illness as biographical disruption. *Sociology of Health and Illness*, 4(2), pp.167–182.
- Carricaburu, D. & Pierret, J., 1995. From biographical disruption to biographical reinforcement: The case of HIV-positive men. *Sociology of Health & Illness*, 17(1), pp.65–88.
- Damar, A.P. & du Plessis, G., 2010. Coping Versus Grieving in a “Death-accepting” Society: AIDS-Bereaved Women Living with HIV in Indonesia. *Journal of Asian and African Studies*, 45(4), pp.424–431.
- Ellis, C., Adams, T.E. & Bochner, A.P., 2011. Autoethnography: An Overview. *Historical Social Research-Historische Sozialforschung*, 12(1), pp.273–290.
- Featherstone, K. et al., 2005. *Risky relations: family, kinship, and the new genetics*, Berg Publishers.
- Greer, G., 2007. *The Whole Woman*, London: Black Swan.

- Grinyer, A., 2007. The biographical impact of teenage and adolescent cancer. *Chronic illness*, 3(4), pp.265–277.
- Hazan, F. et al., 2013. A novel missense mutation of the paired box 3 gene in a Turkish family with Waardenburg syndrome type 1. *Molecular Vision*, 19.
- Heritage, J. & Robinson, J.D., 2006. Accounting for the visit: giving reasons for seeking medical care. In J. Heritage & D. W. Maynard, eds. *Communication in medical care interaction between primary care physicians and patients*. Cambridge, UK: Cambridge University Press, pp. 48–85.
- Homma, M. et al., 2016. “This really explains my case!”: biographical reconstruction of Japanese people with fibromyalgia meeting peers. *Health Sociology Review*, 25(1), pp.62–77.
- Jalilian, N. et al., 2015. A novel mutation in the PAX3 gene causes Waardenburg syndrome type I in an Iranian family. *International Journal of Pediatric Otorhinolaryngology*, 79(10).
- Jiwa, M. et al., 2010. How do Specialist Breast Nurses help breast cancer patients at follow-up? *Collegian*, 17(3), pp.143–149.
- Kearney, P.J., 2006. Autopathography and humane medicine: The diving bell and the butterfly--an interpretation. *Medical humanities*, 32(2), pp.111–113.
- Khan, A., 2007. *Waardenburg syndrome: A volume in the Genetics and Communication Disorder Series*, San Diego; Oxford; Brisbane: Plural Publishing.
- Lattig, M.C. et al., 2008. Deafness on the island of Providencia - Colombia: different etiology, different genetic counseling. *Genetic counseling (Geneva, Switzerland)*,

19(4), pp.403–412.

Locock, L., Ziebland, S. & Dumelow, C., 2009. Biographical disruption, abruption and repair in the context of motor neurone disease. *Sociology of health & illness*, 31(7), pp.1043–1058.

Mousty, E. et al., 2015. A homozygous PAX3 mutation leading to severe presentation of Waardenburg syndrome with a prenatal diagnosis. *Prenatal Diagnosis*, 35(13), pp.1379-1381.

Partington, M.W., 1959. An english family with waardenburg's syndrome. *Archives of Disease in Childhood*, 34(174), pp.154-157.

Read, A.P. & Newton, V.E., 1997. Waardenburg syndrome. *Journal of medical genetics*, 34(8), pp.656–665.

Reeve, J. et al., 2010. Revisiting biographical disruption: exploring individual embodied illness experience in people with terminal cancer. *Health*, 14(2), pp.178–195.

Sharma, K. & Arora, A., 2015. Waardenburg Syndrome: A Case Study of Two Patients. *Indian Journal of Otolaryngology and Head & Neck Surgery*, 67(3), pp.324–328.

Shprintzen, R.J., 2007. Foreword. In *Waardenburg Syndrome: A volume in the genetic syndromes and communication disorders series*. San Diego; Oxford; Brisbane: Plural Publishing.

Song, J. et al., 2016. Hearing loss in Waardenburg syndrome: A systematic review. *Clinical Genetics*, 89(4), pp.416-425).

- Sparkes, A.C., 2017. Autoethnography comes of age: Consequences, comforts, and concerns. In D. Beach, C. Bagly, & S. Marques da Silva, eds. *Handbook of Ethnography of Education*. London: Wiley. In Press
- Sveen, U., Sørberg, H.L. & Østensjø, S., 2016. Biographical disruption, adjustment and reconstruction of everyday occupations and work participation after mild traumatic brain injury. A focus group study. *Disability and rehabilitation*, 38(23) pp.2296–2304.
- Tassabehji, M. et al., 1993. Mutations in the PAX3 gene causing Waardenburg syndrome type 1 and type 2. *Nature Genetics*, 3(1), pp.26–30.
- Waardenburg, P.J., 1951. A new syndrome combining developmental anomalies of the eyelids, eyebrows and nose root with pigmentary defects of the iris and head hair and with congenital deafness. *American journal of human genetics*, 3(3), pp.195–253.
- Wheeler, S.L., 2012. Singing hands in Llangollen! *Sara Louise Wheeler*. Available at: <https://saralouisewheeler.wordpress.com/2012/09/26/singing-hands-in-llangollen/> [Accessed December 31, 2016].
- Wheeler, S.L. & Hopwood, A.G., 2015. Tinnitus: A Deafhearing Phenomenon. *Qualitative Inquiry*, 21(2), pp.173–174.
- Wikipedia, 2016. Waardenburg syndrome. *Wikipedia: the free encyclopedia*. Available at: https://en.wikipedia.org/wiki/Waardenburg_syndrome [Accessed December 23, 2016].
- Wilson, S., 2007. "When you have children, you're obliged to live": Motherhood,

chronic illness and biographical disruption. *Sociology of health & illness*, 29(4), pp.610–626.

Withey, R.-A., 2016. Why “invisible” hearing aids are not for me. Hearing aids need more visibility. *The Limping Chicken*. Available at: <http://limpingchicken.com/2016/11/25/rebecca-anne-withey-why-invisible-hearing-aids-are-not-for-me-hearing-aids-need-more-visibility/> [Accessed December 31, 2016].

Wright Mills, C., 1959. *The sociological imagination*, New York: Oxford University Press.

Xiao, Y. et al., 2016. A novel mutation in PAX3 associated with Waardenburg syndrome type I in a Chinese family. *Acta Oto-Laryngologica*, 136(5), 439-445.

Yoshida, K.K., 1993. Reshaping of self: a pendular reconstruction of self and identity among adults with traumatic spinal cord injury. *Sociology of Health and Illness*, 15(2), pp.217–245.